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Letter to the Editor

Verbal-Performance Discrepancies in a Family With Noonan Syndrome

To the Editor:

INTRODUCTION

Noonan syndrome affects approximately 1 per 1,000 individuals, although estimates of its prevalence have ranged from 1 per 100 in those mildly affected to 1 per 2,500 in those severely affected [Nora and Fraser, 1981; Nora et al., 1974]. Until recently, the gene for Noonan syndrome had not been mapped, although the high number of familial cases with transmission from mother to children had indicated the possible role of X-linked dominant genes in the transmission of the disorder [Mendez and Opitz, 1985]. However, the result of a linkage analysis of a three-generation family with eight affected individuals and 20 two-generation families by Jamieson et al. [1994] localised a gene for Noonan syndrome to chromosome region 12q22-qter.

A distinctive cognitive profile of verbal fluency and good vocabulary in contrast to poor non-verbal skills (e.g.,visuo-constructional skill) [McCauley et al., 1987; Nyborg, 1990] was reported in a group of eight males with NS [Money and Kalus, 1979], although close examination of the pattern of results shows considerable variability of performance. Other cognitive traits of NS include delayed motor development [Lemmi et al., 1983], learning disability, articulation abnormalities [Allanson, 1987], and autism [Ghaziuddin et al., 1994].

Given the frequency of NS and its potential contribution to an understanding of cerebral lateralisation of function, it is surprising that relatively few studies have examined cognitive abilities in affected males and females.

In the present report, the cognitive profiles of three relatives (mother and two daughters) with NS are described to demonstrate a possible pattern of strengths and weaknesses within this syndrome.

METHOD AND RESULTS

Three female relatives all diagnosed as having NS participated in the study; HB (mother 24.6 years), LB (daughter 5.1 years), and CB (daughter 4.2 years).

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HB

Verbal and performance skills were assessed using the Wechsler Adult Intelligence Scale-Revised (WAIS) [Wechsler, 1986]. This documented a Verbal IQ of 77 (borderline) and a Performance IQ of 88 (low average). The difference in scores between these two scales was significant at the 0.05% level of significance. Lowest scores were obtained on the Comprehension, Similarities, and Information subscales of the Verbal scale, while the highest scores were obtained in the Block Design, Object Assembly and Digit Symbol subscales of the Performance scale. In the former tests, the ability to comprehend and store verbal material is required while the latter tasks require constructional manipulation of spatial designs.

LB and CB

Both LB and CB were assessed on the Merrill-Palmer Scale of Mental Tests [Stutsman, 1948] which is designed specifically for pre-school children. While it contains some basic language tests (comprehension, verbal repetition, and general knowledge), it predominantly measures performance abilities incorporating such tests as all-or-none tests (e.g., throwing a ball, building a tower, or standing on one foot), formboards and picture tests, and tests of motor co-ordination (e.g., pegboards and building pyramids). For this reason, the scale has been used extensively with young autistic children as a means of measuring ability [Howlin, 1991] and because only minimal language skill is needed to complete the test, it allows children to be rated by it when a rating would be impossible on other standardised tests such as the Stanford Binet test and the Wechsler Intelligence Scale for Children (WISC). Scores on this scale revealed a mental age (MA) almost equivalent to chronological age (CA) for both sibs: LB = 4 years 8 months and CB = 4 years 0 months. However, when the language tests are examined separately from the non-verbal tests, both LB and CB performed worse on the language test (in particular, comprehension and general knowledge) scoring at a level of below 3 years compared to their performance on non-verbal tasks (in particular, the Seguin formboard and picture puzzles), scoring at a level above 5 years.

In order to examine language abilities in greater detail, LB and CB were assessed on the Reynell Developmental Language Scale (RDLS) [Reynell, 1977] which measures verbal comprehension and expressive language and the British Picture Vocabulary Scale (BPVS) [Dunn et al., 1982] which measures receptive vocabulary. LB

scored at an age equivalent of 3 years 10 months on the comprehension scale and 2 years 9 months on the expressive language scale of the RDLS. Her score on the BPVS was equivalent to 3 years 0 months. This pattern was repeated in CB who scored the age equivalent of 3 years 0 months on the comprehension scale and 2 years 10 months on the expressive scale of the RDLS. Her score on the BPVS was equivalent to 2 years and 3 months.

DISCUSSION

The results of the present study suggest a pattern of cognitive strengths and weaknesses in three relatives with Noonan syndrome. Mother and daughters appear to have strengths in non-verbal skills, particularly those requiring manipulations of spatial designs, but weakness in verbal skills such as those requiring comprehension and retention of verbal information. This finding contrasts with that reported by Money and Kalus [1979], who found verbal ability to be a relative strength in comparison to performance ability in a group of males with NS. However, the verbal deficits reported in the present study are in accordance with the findings of Wilson and Dyson [1982] who reported delayed comprehension and receptive language in a 7-year-old girl with NS. Unfortunately, their assessment did not include non-verbal performance.

The present finding of a verbal-performance discrepancy in the direction of superior performance skill compared to verbal skill may well be indicative of possible anomalous cerebral lateralisation in NS. However, the extent to which this dissociation in cognitive performance differs between males and females is in need of further clarification. If males show superior verbal skill in comparison to their performance skill and if females show the reverse pattern with verbal skill superior to performance skill then it may indicate differing genetic influences in the cognitive performance of males and females with NS.

A potentially illuminating finding of the present study is the homogeneity in the pattern of cognitive performance in a family with NS. This suggests that the often reported variability in the clinical phenotype of familial cases [Mendez and Opitz, 1985] may not extend to include the behavior phenotype. It has been suggested that the wide clinical differences seen within families with NS may preclude recognition and hence diagnosis of the adult gene carrier [Allanson, 1985]. However, defining the cognitive profile of families with NS may help enhance the order of selection of screening in those individuals whose clinical findings are very subtle. It will also aid in the establishment of more appropriate genetic counselling for both affected individuals and their families.

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